



ETFA gene

electron transfer flavoprotein alpha subunit

Normal Function

The *ETFA* gene provides instructions for making one part (the alpha subunit) of an enzyme called electron transfer flavoprotein. This enzyme is normally active in the mitochondria, the energy-producing centers in cells. Electron transfer flavoprotein is involved in the process by which fats and proteins are broken down to produce energy.

Health Conditions Related to Genetic Changes

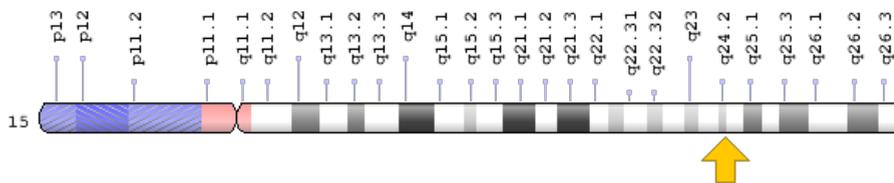
glutaric acidemia type II

Some mutations in the *ETFA* gene prevent the production of the electron transfer flavoprotein enzyme. Other mutations result in the production of a defective enzyme that cannot fulfill its role in the series of reactions (metabolic pathways) that break down fats and proteins. This enzyme deficiency allows these nutrients, as well as compounds created as the nutrients are partially broken down, to build up to abnormal levels, especially when the body is under stress. Toxic products of incomplete metabolism damage cells in many body systems, resulting in the signs and symptoms of glutaric acidemia type II.

Chromosomal Location

Cytogenetic Location: 15q24.2-q24.3, which is the long (q) arm of chromosome 15 between positions 24.2 and 24.3

Molecular Location: base pairs 76,216,228 to 76,311,469 on chromosome 15 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- electron transfer flavoprotein alpha-subunit
- electron-transfer-flavoprotein, alpha polypeptide
- Electron transfer flavoprotein, alpha polypeptide
- electron-transfer-flavoprotein, alpha polypeptide (glutaric aciduria II)
- electron-transferring-flavoprotein, alpha polypeptide (glutaric aciduria II)
- EMA
- ETFA_HUMAN
- GA2
- MADD

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ETFA%5BTIAB%5D%29+OR+%28%28MADD%5BTIAB%5D%29+OR+%28electron+transfer+flavoprotein+alpha-subunit%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

OMIM

- ELECTRON TRANSFER FLAVOPROTEIN, ALPHA POLYPEPTIDE
<http://omim.org/entry/608053>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_ETFA.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ETFA%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3481
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/2108>
- UniProt
<http://www.uniprot.org/uniprot/P13804>

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